AGSD UK
Association for Glycogen Storage Disease

INTRODUCTION

Your Pompe resources

These information sheets have been designed with community members and other experts to help with the different challenges that living with Pompe can bring.

You can choose and click on the sheets that are relevant to you or let us know if you'd like us to send hard copies, plus a folder for storing your Pompe information.

We'll continue to add new sheets to the series - if there's a topic you want to see covered just let us know.

Whatever issues you face living with Pompe, AGSD-UK is here to help. Our offer includes specialist care and advisory services and a range of groups and activities to bring people together and help with living well.

"Pompe has a massive impact on our lives but we have found a way to integrate it as just another little part of who we are."

Parents of children with infant onset Pompe

AGSD-UK greatly appreciates the financial assistance of our sponsors with our range of publications for people affected by glycogen storage disorders.

For all queries and support contact: info@agsd.org.uk

About Pompe

Pompe is a rare, progressive condition that causes muscle weakness.

It's also known as Glycogen Storage Disease type II or GSD2.

The condition is inherited from parents who are carriers. It's caused by mutations in a gene that makes an enzyme called acid alpha-glucosidase (GAA), which the body uses to break down glycogen - a stored form of sugar used for energy.

The absence or reduced level of this enzyme in people affected by Pompe means that glycogen builds up in muscle cells and causes damage.

There are two types of Pompe: infantile onset and late onset.

Infantile onset is the more severe form and babies will often show heart and respiratory symptoms within the first few weeks after birth. A variant form of infantile onset associated with less severe heart problems usually presents in the first year of life.

In late onset, symptoms can develop in children, teenagers or adults and can be mild or more severe. They may include problems with movement such as walking, climbing stairs or getting up from a chair, pain, headache and fatigue. These symptoms may be quite subtle at first but can make everyday activities more challenging.

There are currently three types of enzyme replacement therapy available in the UK to help with the symptoms of Pompe. There's also ongoing research into gene therapy and other forms of treatment.

"My Pompe diagnosis was not completely confirmed until I got my genetics report...by this time the news wasn't a shock, I was expecting it, but it hit home hard seeing it in black and white...I am now receiving fortnightly infusions at home from a fantastic nurse.

AGSD-UK have supported me so much over the last months... My Pompe journey is only in its infancy but ...I am looking forward to a positive future with Pompe and me"

Person with late onset Pompe

If you, your child or someone close to you has recently been diagnosed with Pompe it can be hard to know where to turn.

Visit the Pompe pages on our website agsd.org.uk for more information and signposting to further support and register with us to keep updated on community events.

Remember

AGSD-UK's advice and support services are here for you and we can help put you in touch with others who've been through similar experiences. Just contact **info@agsd.org.uk**